

## Mannose-binding lectin deficiency

### Description

Mannose-binding lectin deficiency is a condition that affects the immune system. People with this condition have low levels (deficiency) of an immune system protein called mannose-binding lectin in their blood. Whether this deficiency makes affected individuals prone to recurrent infections is not clear.

People with mannose-binding lectin deficiency can develop infections of the upper respiratory tract and other body systems. Individuals with this condition may also contract more serious infections such as pneumonia and meningitis. Depending on the type of infection, the symptoms caused by the infections vary in frequency and severity.

Infants and young children with mannose-binding lectin deficiency seem to be more susceptible to infections than affected adults, but adults can also develop recurrent infections. In addition, affected individuals undergoing chemotherapy or taking drugs that suppress the immune system are especially prone to infections.

### Frequency

Mannose-binding lectin deficiency is thought to affect approximately 5 to 10 percent of people worldwide; however, many affected individuals have no signs or symptoms related to low mannose-binding lectin levels. The condition is more common in certain populations, such as sub-Saharan Africans.

### Causes

Relatively common mutations in the *MBL2* gene can lead to mannose-binding lectin deficiency. This gene provides instructions for making a protein that assembles into a complex called mannose-binding lectin. Functional mannose-binding lectins are made up of two to six protein groups called trimers, which are each composed of three of the protein pieces (subunits) produced from the *MBL2* gene.

Mannose-binding lectin plays an important role in the body's immune response by attaching to foreign invaders such as bacteria, viruses, or yeast and turning on (activating) the complement system. The complement system is a group of immune system proteins that work together to destroy foreign invaders (pathogens), trigger inflammation, and remove debris from cells and tissues. Mannose-binding lectin can also stimulate special immune cells to engulf and break down the attached pathogen.

Mutations in the *MBL2* gene can reduce the production of the mannose-binding lectin subunit or eliminate the subunit's ability to assemble into functional mannose-binding lectin. A decrease in the availability of the normal subunit protein may lead to a reduction of the functional mannose-binding lectin in blood. With decreased levels of mannose-binding lectin, the body does not recognize and fight foreign invaders efficiently. Consequently, infections can be more common in people with this condition.

However, not everyone with a change in the *MBL2* gene has decreased levels of mannose-binding lectin, and not everyone with decreased protein levels is prone to infection. Researchers believe that a number of factors, including other genetic and environmental factors, are involved in the development of mannose-binding lectin deficiency and susceptibility to infection.

[Learn more about the gene associated with Mannose-binding lectin deficiency](#)

- MBL2

## **Inheritance**

The inheritance pattern of mannose-binding lectin deficiency is unclear. Some reports show that having a disease-associated mutation in one copy of the *MBL2* gene in each cell can lead to the condition, while other reports state that a mutation in both copies of the gene is necessary. It is important to note that people inherit an increased risk of developing mannose-binding lectin deficiency, not the condition itself. Not all people who inherit mutations in this gene will develop the condition.

## **Other Names for This Condition**

- Mannose-binding lectin protein deficiency
- Mannose-binding protein deficiency
- MBL deficiency
- MBL2 deficiency
- MBP deficiency

## **Additional Information & Resources**

### Genetic Testing Information

- Genetic Testing Registry: Mannose-binding protein deficiency (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3280586/>)

### Genetic and Rare Diseases Information Center

- Mannose-binding lectin protein deficiency (<https://rarediseases.info.nih.gov/disease>)

s/10309/mannose-binding-lectin-protein-deficiency)

#### Patient Support and Advocacy Resources

- Disease InfoSearch (<https://www.diseaseinfosearch.org/>)
- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

#### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov (<https://clinicaltrials.gov/ct2/results?cond=%22mannose-binding+lectin+deficiency%22>)

#### Catalog of Genes and Diseases from OMIM

- MANNOSE-BINDING LECTIN DEFICIENCY (<https://omim.org/entry/614372>)

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28mannose-binding+lectin+deficiency%5BTIAB%5D%29+OR+%28mannose-binding+protein+deficiency%5BTIAB%5D%29+OR+%28mbl+deficiency%5BTIAB%5D%29+OR+%28mbl2+deficiency%5BTIAB%5D%29+OR+%28mbp+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>)

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**Last updated May 1, 2018**